

Case Report

Polycystic kidney and Down Syndrome

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Abstract

Kidney disease is not a common complication in Down Syndrome (DS). A variety of renal and urologic problems have been described in these patients and some develop renal failure. Coincidence of polycystic kidney disease and DS is a rare entity. This is a report of a 3.5-month-old infant with DS, known to have early end-stage renal failure due to polycystic kidney disease.

JRMS 2006; 11(4): 278-280

Down syndrome (DS) is the most common chromosomal abnormality with multiple-organ involvement. The incidence of renal and urologic anomalies has been estimated at 3.5-21.5%¹. Some authors have suggested that the incidence of these anomalies is high enough to encourage systemic screening of these patients². Also, due to increased survival the number of these patients with renal failure is growing^{1,3}. Therefore, regular monitoring of patients with DS for renal and urinary tract disorders from early infancy to adulthood has been recommended^{1,3,4}.

Case Report

A 3.5-month-old infant was admitted for fever, poor feeding and anuria. He was the third child of healthy unrelated parents (mother's age was 35 and father's age was 40, with normal renal ultrasonography). There was no history of renal disease in their families. The patient had bilateral renal enlargement with severe hyperechogenicity in his prenatal period. Physical examination revealed all the hallmarks of DS, which was previously confirmed by chromosomal analysis. BP was 109/98 mm/Hg (on 95% standard curve). Both kidneys were palpable and enlarged. Laboratory exams upon admission were as follows: Hb 9 g/dl, serum Na 128 meq/L and K 4.5 meq/L, Ca 8 mg/dl, P 12.7 mg/dl, creatinine 5.7

mg/dl and BUN 68 mg/dl. He also had metabolic acidosis. A specific gravity of 1006, protein +, sugar ++, 10-15 RBCs, many WBCs and WBC clumps were reported in urinalysis. Urine culture was negative. In abdominal ultrasound, both kidneys were enlarged with marked hyperechogenicity, suggestive of infantile polycystic kidney disease (figures 1 and 2). In abdominal CT scan, both kidneys demonstrated a symmetric and smooth enlargement with low density of both kidneys, secondary to accumulation of urine in dilated renal tubules as an expected finding in the context of autosomal recessive polycystic kidney disease. It was impossible to use contrast material for IVP or to perform DMSA scan due to severe renal failure in this patient. Parents refused renal biopsy in their child. Peritoneal dialysis and antibiotic therapy were started for the treatment of renal failure, but serum creatinine stabilized at about 3.5 mg/dl in the following days. Therefore, continuous ambulatory peritoneal dialysis (CAPD) was started for the established end-stage renal disease in this patient.

Discussion

We present the case of a patient with DS with marked bilateral hyperechogenic enlarged kidneys since the prenatal period. Bilateral

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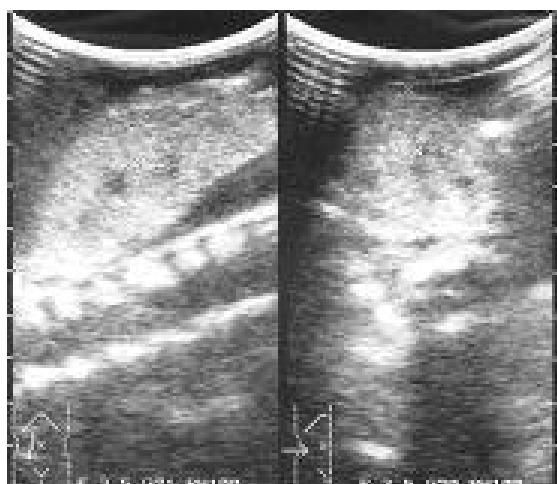


Figure 1. Abdominal ultrasound.

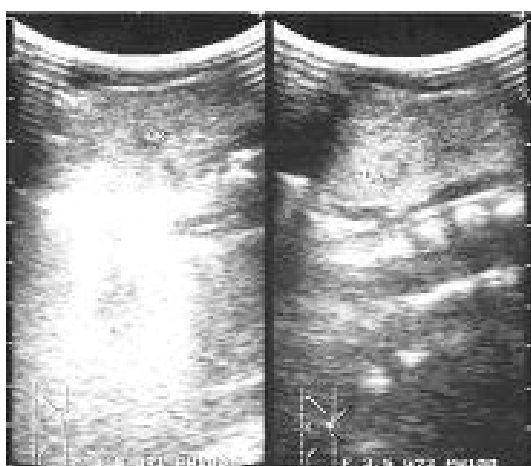


Figure 2. Abdominal ultrasound.

enlarged kidneys can be found in any proliferative disorder, abnormal deposition of proteins such as amyloidosis and multiple myeloma, tumoral cell proliferation such as lymphoma and leukemia, glycogen storage disease, acute tubular necrosis, late phase of acute cortical necrosis, acute interstitial nephritis, glomerulocystic kidney, homozygote sickle cell anemia, liver cirrhosis, Beckwith Wiedemann syndrome, Bartter syndrome, Fabry disease, total parenteral nutrition, lasix and vasodilators prescription, hemophilia, and paroxysmal nocturnal hemoglobinuria⁵. None of the above was consistent with history, laboratory findings and radiologic exams in this case. Bilateral hyperechogenic kidneys are also seen in autosomal recessive polycystic kidney (ARPKD)

and sometimes in autosomal dominant polycystic kidney (ADPKD)⁵.

In 1960, Berg described renal agenesis, hypoplasia and horseshoe kidney in Down Syndrome⁶. In a study of 124 autopsy cases with DS, Atrial described anomalies such as renal hypoplasia, tubular dilation, glomerular microcysts, simple cysts, immature glomeruli in deep cortex, obstructive uropathies such as ureterovesical junction obstruction, ureteral stenosis, and bilateral cystic dysplastic kidneys. He suggested chromosomal analysis in any case with prenatal or neonatal obstructive renal lesions⁷.

Kupferrman noticed an increased risk of obstructive uropathies in these patients and suggested early screening with ultrasound for kidney and urinary tract abnormalities as part of standard clinical care in these children, and if abnormal, a voiding cystourethrography should be performed⁶. Other urologic problems such as: ureteropelvic junction obstruction¹, posterior urethral valve⁸, ectopic kidney⁹, mild fetal pyelectasis¹⁰, reflux nephropathy, voiding dysfunction¹ Hinman syndrome¹¹, and renal pathologies such as focal segmental glomerular sclerosis, acute crescentic glomerulonephritis, minimal change glomerular lesions, membrane glomerulopathy, deep cortical immature glomeruli, cytomegalovirus infection in renal medulla³, acute tubulointerstitial nephritis¹², renal tubular dysgenesis as insufficient proximal tubules¹³, nephrogenic diabetes mellitus with mutation in V2 receptor¹⁴, and IgG-associated mesangial glomerulonephritis¹⁵ have been reported in this anomaly.

Also, some reports of intestinal absorptive hypercalciuria¹⁶, cystinuria⁴, uricosuria¹, hemolytic uremic syndrome¹⁷, membrane glomerulonephritis⁴ and chronic glomerulonephritis¹⁸ have been published in these patients.

The child presented in this study is a case of PKD with DS that seems to be the first report of this association in our literature review. In ARPKD, kidneys are large and hyperechoic without definite corticomedullary differentia-

tion (CMD). Diffuse echogenic loci are common in end-stage renal failure. Ultrasonographic findings in ADPCK consist of renal cysts and normal to hyperechogenic enlarged kidneys without CMD. These features could be present even in fetal and neonatal life in ADPCK with severe presentation such as ARPCK, and differential diagnosis needs renal biopsy^{19,20}. The gene of ARPCK has been local-

ized to the chromosomal region 6p21-cen²¹. Also, the most common cause of DS is trisomy of chromosome 21. The occurrence of DS and PCKD in this patient could be a coincidental finding, but the common involvement of chromosome 21 in these two diseases is an important finding, which remains to be substantiated by further reports.

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